Correlation of Ultrasonography and MRI in Prenatal Diagnosis of Lobar Holoprosencephaly

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Prenatal diagnosis for alobar holoprosencephaly, is not difficult while for semilobar and lobar forms itmay be difficult. We present and discuss prenatal ultrasonographic and magnetic resonance imaging features of lobar holoprosencephaly in a 29 weeks-old fetus diagnosed prenatally.
A nineteen years-old primigravid woman was referred with diagnosis of hydrocephalus. In ultrasonographic examination, lateral ventricles and cisterna magna were dilated, but absence of cavum septum pellucidum excluded the diagnosis of hydrocephalus. B mode examination of coronal and transverse sections of fetal brain revealed a cystic cavity of partly fused frontal horns between the two hemispheres in the anterior part of skull, and thalamic fusion. However, frontal horns were developed and discernible. Midline echo, lateral ventricles and third ventricle were observed. It was not possible to examine sagital planes of fetal brain by ultrasonography. Ultrasonographic diagnosis was lobar holoprosencephaly, which was then confirmed by fetal MRI. Ultrasonographic and MRI correlation was assessed.
A detailed ultrasonographic evaluation is the primary examination method of obstetrician and it may help to diagnose and grade holoprosencephaly. Besides its correlation with prenatal MRI is good.

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Key Words: Holoprosencephaly, Prenatal diagnosis, Ultrasonography, Magnetic resonance imaging

Introduction

Holoprosencephaly, a genetically heterogeneous disorder resulting from incomplete cleavage of the embryonic forebrain structures occurs in 1:15000 live births.[1] Brain malformations in holoprosencephaly are classified as alobar, semilobar and lobar. Alobar holoprosencephaly is the most severe type with a single midline forebrain ventricle, a cerebral holosphere, completely absent corpus callosum and no interhemispheric suture. The defect is localized more anteriorly with some interhemispheric fissure at the posterior and variable amount of corpus callosum in semilobar form. The mildest form is lobar holoprosencephaly in which cerebral hemispheres are mostly separated with basal frontal aspects non-cleaved.[2,3] Although prenatal diagnosis of holoprosencephaly has been described and not difficult for alobar holoprosencephaly, prenatal diagnosis of semilobar and lobar forms may be difficult.[2,4] Besides, grading of holoprosencephaly is not performed prenatally and correlation of ultrasonography and MRI in holoprosencephaly is not documented. We present and discuss prenatal ultrasonographic and magnetic resonance imaging features of lobar holoprosencephaly in a 29 weeks-old fetus diagnosed prenatally.

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Case Report

A nine-teen year old primigravid woman at 29 gestational weeks was referred to department of obstetrics and gynecology, Yüzüncü Yıl University Faculty of Medicine with diagnosis of hydrocephaly. Ultrasonographic examination and diagnosis were made by Picus (Model 410650, Picus Pie Medical Equipment, ESAOTE-Sp.a Italy) and were recorded to digital versatile disc in live video format. MRI sections and diagnosis were taken independently. After digital photographic documentation of magnetic resonance images by Digital Canon Ixusi (PC1060, Canon Inc, Japan), frames of ultrasonographic video were produced in Windows Media Player Classic. MRI and ultrasonographic frames were than compared.

In ultrasonographic examination, lateral ventricles and cisterna magna were dilated, but absence of cavum septum pellucidum excluded the diagnosis of hydrocephalus. B mode examination of coronal and transverse sections of fetal brain revealed a cystic cavity of partly fused frontal horns between the two hemispheres in the anterior part of skull and thalamic fusion. However, frontal horns were developed and discernible. Midline echo and lateral ventricles and third ventricle were observed. Figure 1. It was not possible to examine sagital planes of fetal brain by ultrasonography. There was no addi-

tional sign of anomaly. Ultrasonographic diagnosis was lobar holoprosencephaly, which was then confirmed by fetal MRI. Frontal horns were discernible and partly fused. There was thalamic fusion and absent cavum septi pellucidum in MRI examination. Cerebral hemispheres were partly fused. Interhemispheric fissure was identifiable. Correlation of ultrasound and MRI was good. However, sections examined in ultrasonography were mainly dependent on fetal presentation and sagital sections were not available.

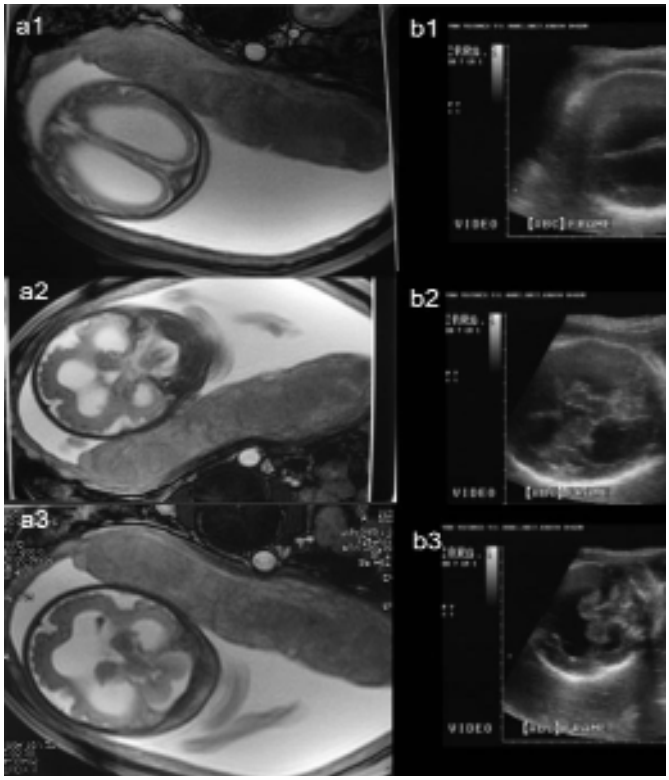

Figure 1: MRI sections (a) and corresponding ultrasonographic sections (b). Midline echo, dilated lateral ventricles and third ventricle are observed (a1,a2,b1,b2). Frontal horns are developed and discernible however, a cystic cavity of partly fused frontal horns between the two hemispheres in the anterior part of skull and thalamic fusion is observed in a3,b3

Laboratory examination of the mother including toxoplasmosis, cytomegalovirus serology was unremarkable. The patient was then followed up and delivered a breech presenting, 2820 gr. male fetus at 36 weeks of gestation by cesarean section. First and fifth minute Apgar were 5 and 7, respectively. Postpartum ultrasonographic examination of the baby was in accordance with intrauterine evaluation. The baby was taken to neonatal care unit as he had respiratory distress syndrome and pulmonary hypertension. He died on postpartum third day. Parents did not give permission for autopsy and karyotyping because of religious believes.

Discussion

Holoprosencephaly develops as a consequence of failure of cerebrum to form distinct left and right hemispheres and is classified according to severity of brain morphology as alobar, semilobar and lobar.[2] Type of holoprosencephaly with associated anomalies determines prognosis of the case.[5]

Mental retardation, hypotelerosim, nasal defects, cleft palate, microcephaly, single central maxillary incisor and congenital nasal pyriform aperture stenosis may occur in association with holoprosencephaly. Presence of proboscis, hypotelerosim and alobar holoprosencephaly is classified as ethmocephaly.[3] It is proposed that face malformation predicts brain malformation.[3,6] Therefore, craniofacial examinations of these cases are ultimately important. But on the other hand, diagnosis and classification of holoprosencephaly is difficult and can not always be precise, even after birth. Some cases can not be classified into one form and prognosis of some cases does not correlate with the classification.[7] Barkovic et al reported that temporal and occipital lobes are well-differentiated in children with holoprosencephaly, however frontal lobes are not.[8] If no ventricles could be identified and no demonstrable interhemispheric fissure was observed, they classified holoprosencephaly as alobar. If the temporal lobes were somewhat differentiated, with some true temporal horn formation in the lateral ventricles, or if some true interhemispheric fissure was observed, the holoprosencephaly was classified as semilobar. If the lateral ventricles were adequately differentiated such that some frontal horn formation was observed, the holoprosencephaly was classified as lobar. They further subdivided into severe semilobar and mild semilobar. If a true lateral ventricular body was formed and was observed to be extending superolaterally and anterior to the thalamus, the semilobar holoprosencephaly was classified as mild. If the only portion of the lateral ventricle that was differentiated was the temporal horn, the semilobar holoprosencephaly was classified as severe.

Prenatal diagnosis is not always precise and there are no efforts to grade the anomaly. We present ultrasonographic and MRI characteristics of lobar holoprosencephaly in order to classify and grade the anomaly. Besides, we observed the correlation of prenatal MRI and ultrasonography in holoprosencephaly. Prenatal ultrasonographic examination was in accordance with prenatal MRI. In our case, temporal lobes were developed. Frontal horns were developed and discernible except in one section. Cavum septum pellucidum was absent and there was thalamic fusion, both in ultrasonographic and MRI images. It was difficult to distinguish between mild holoprosencephaly and lobar holoprosencephaly. Whether it was mild semilobar holoprosencephaly or lobar holoprosencephaly, we expected a better prognosis than severe semilobar and alobar holoprosencephaly. But, the baby died soon after birth in our case.

Holoprosencephaly may occur in association with chro-

mosomal abnormalities, particularly trisomy 13. It may also be associated with autosomal recessive disease such as Meckel-Gruber syndrome. Therefore, targeted genetic ultrasonography and detailed examination of the other systems is utmost important. Karyotyping of fetus may be offered to the patient in early gestational weeks.

In ultrasonographic evaluation it is important to examine the all available planes and to examine as many as of the possible sections in each plane. MRI can help to confirm the diagnosis and grade of holoprosencephaly, but a detailed ultrasonographic evaluation is the primary examination method of obstetrician and it may help to diagnose and grade holoprosencephaly. Besides its correlation with prenatal MRI is good. Every effort to classify and grade holoprosencephaly should be undertaken prenatally.

Lobar Holoprosensefalinin Prenatal Tanısında Ultrasonografi ve Magnetic Rezonans Görüntülemenin Korelasyonu

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Alobar holoprosensefalinin (HPE) prenatal tanısı kolayca yapılabilmekteysede, semilobar ve lobar formların teşhisi güçtür. Çalışmamızda 29 haftalık bir fetusta prenatal ultrasonografi ve magnetic rezonans görüntülemesiyle tanı konulmuş bir lobar HPE'li fetusun bulguları sunulmuş ve tartışılmıştır. Ondokuz yaşında primigravid bir hasta hidrosefali tanısıyla kliniğimize refere edildi. Ultrasonografik muayenede lateral ventrikül ve sisterna magna dilate izlendi, fakat kavum septum pellusidumun olmaması hidrosefali tanısını ekarte ettirdi. Koronal ve transvers B mod ultrasonografik kesitlerde her iki hemiküre arasında frontal boynuzların birleşerek yaptığı kistik bir genişleme ve thalamik füzyon izlenmişti ancak frontal boynuzlar ayrı bir yapı olarak gözlemlenmekteydi. Orta hat eko, lateral ventrikül ve 3. ventrikül izlenebilmekteydi. Ultrasonographik olarak sagital kesit almak mümkün olmamıştı. Ultrasonografik olarak konulan lobar HPE tanısı daha sonar MRI ile doğrulandı. HPE tanısında ve derecelendirilmesinde esas muayene yöntemi ultrasonografik yöntemdir. Ultrasonografinin MRI ile korelasyonu iyi derecededir.

Anahtar Kelimeler: Holoprosensefali, Prenatal tanı, Ultrasonografi, Magnetic rezonans görüntüleme

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